
DISEASE:
Craniofonticulusutural dysplasia

NAME:	Craniofonticulusutural dysplasia
DESCRIPTION:	Craniofonticulusutural dysplasia (CLSD), also known as Boyadjiev-Jabs syndrome, is characterized by the specific association of large and late-closing fontanelles, hypertelorism, early-onset cataract and mild generalized skeletal dysplasia.
ORPHACODE:	50814
SYNONYMS:	Boyadjiev-Jabs syndrome
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>SEC23A</u>
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